ABSTRACT

A novel photoreceptor/pineal-expressed gene encoding aryl-hydrocarbon receptor interacting protein-like 1 (*AIPL1*), the associated protein like amino acid sequence and methods for identifying the presence of the sequence in patients. Leber congenital amaurosis (LCA) is the most severe form of inherited retinal dystrophy and the most frequent cause of inherited blindness in children. LCA is usually inherited in an autosomal recessive fashion, although rare dominant cases have been reported. One form of LCA, LCA4, maps to chromosome 17p13 and is genetically distinct from other forms of LCA. The inventors recently identified the gene associated with LCA4, *AIPL1* (aryl-hydrocarbon receptor interacting protein-like 1) and identified three mutations that were the cause of blindness in five families with LCA.